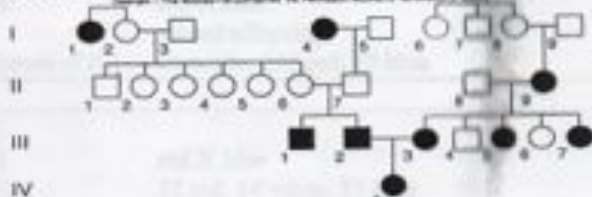


I. Multiple Choice Section of the Test. Place the letter of the most correct answer to each question on the provided Answer Sheet. Each correct answer is worth two points. There is no penalty for guessing.

1. The pedigree shown below indicates the occurrence of an unusually frequent, mild genetic disease (the symptoms are not very severe). Assume the trait is fully penetrant. What is the best description of what you can determine about the mode of inheritance of the disease allele.



- A) The disease allele is definitely recessive and likely X-linked.
 B) The disease allele is definitely dominant and likely X-linked.
 C) The disease allele is definitely recessive and likely autosomal.
 D) The disease allele is definitely dominant and likely autosomal.
 E) The disease allele is definitely recessive and definitely autosomal.

2. A term that expresses the degree to which the phenotype is expressed in an individual with the genotype associated with that phenotype.

- A) Penetrance
 B) Pleiotropy
 C) Expressivity
 D) Phenocopy
 E) Epistasis

3. Two strains of pure breeding, deaf mice have been isolated. The Strain 1 deafness allele is recessive to the wild type allele and is autosomal. Similarly the Strain 2 deafness allele is recessive to the wild type allele and is autosomal. There is no evidence of linkage. Strain 1 females are mated with Strain 2 males. All of the female and male F_1 offspring have wild type quality hearing. The reciprocal cross gives the same result- all F_1 offspring have wild type quality hearing. When several F_1 offspring are mated with each other to yield an F_2 , what is the most likely expected ratio for hearing in the offspring?

- A) 9 wild type hearing, 7 deaf
 B) 3 deaf, 1 wild type hearing
 C) 1 deaf, 2 poor hearing: 1 wild type hearing
 D) 3 wild type hearing, 1 deaf
 E) 1 deaf, 1 wild type hearing

4. A couple are both heterozygous for two, independently assorting, autosomal recessive diseases: cystic fibrosis (CF) and phenylketonuria (PKU). What is the probability that their first child will have either CF or PKU, but not both?

- A) 1/16
 B) 6/16
 C) 7/16
 D) 8/16
 E) 9/16

Version 1

9:3:3:1

CP
 CP
 CF

$FfKk \times FfKk$

Strain
 cf. $aaBB \times AAbb$
 $fAaBb$

page 2

5. In a certain plant species flower color may be red, white or blue. A series of crosses was made to determine the mode of inheritance of flower color in this species. Three separate crosses, the results of which are given below, were performed using parents that may or may not have been homozygous.

Cross 1

Parental	red X blue
F ₁	all red offspring
F ₂ (selfcross of F ₁)	61 white, 236 red, 19 blue

Cross 2

Parental	red X blue
F ₁	32 red, 14 white, 17 blue

Cross 3

Parental	red X white
F ₁	12 blue, 45 red, 35 white

The genotype of the red parent in Cross 2 is

- A) AA
- B) AABB
- C) AaBb
- D) aabb
- E) aaBB or AAbb

Handwritten notes:
 32 red
 14 white
 17 blue
 ∴ Red is dominant
 Rr Pp

6. All of the following cells, shown in various stages of mitosis or meiosis, come from the same rare species of plant.



What is the haploid chromosome number in this plant and which stage represents a stage of mitosis?

- A) haploid number is 3 and stage X is mitotic
- B) haploid number is 3 and stage Y is mitotic
- C) haploid number is 3 and stage Z is mitotic
- D) haploid number is 6 and stage X is mitotic
- E) haploid number is 6 and stage Y is mitotic

Handwritten notes:
 Alls are
 Diploid
 n for same

7. Synapsis of homologous chromosomes occurs during

- A) the second meiotic division.
- B) the zygotene stage of prophase I.
- C) prophase II.
- D) diplotene.
- E) anaphase I

12. In corn plants smooth leaves are dominant to crinkled ones; for a second trait tall plants are dominant to dwarf plants. A true breeding line of tall plants with crinkly leaves is crossed to another true breeding line of dwarfed plants with smooth leaves. The resulting F_1 is self-crossed and the resulting F_2 have:

104 tall plants with smooth leaves
 43 dwarf plants with smooth leaves
 51 tall plants with crinkled leaves
2 dwarf plants with crinkled leaves
 200 total

The two genes being studied

- A) are not linked
 B) are linked at 20 cM
 C) are linked at 2 cM
 D) are linked at 1 cM
 E) are linked at 10 cM

13. You observe the following: i) a small loop in polytene chromosomes, and
 ii) reduced genetic map for the affected chromosome and
 iii) pseudodominance of three loci

The most likely cause is

- A) heterozygosity for a duplication.
 B) heterozygosity for a deletion.
 C) heterozygosity for an inversion.
 D) homozygosity for a duplication.
 E) Two of the provided answers are equally correct.

14. Acentric fragments are produced in

- A) duplications
 B) paracentric inversions
 C) pericentric inversions
 D) translocations
 E) deletions

15. The role of the chemical Aroclor in the Ames test is to

- A) Stimulate Salmonella's repair systems
 B) Knock out Salmonella's repair systems
 C) Stimulate the rat liver enzymes
 D) Knock out the rat liver enzymes
 E) block histidine biosynthesis

16. You observe a trivalent (three chromosomes tied together by chiasmata) at metaphase I meiosis.

This could be due to

- A) Autotriploidy
 B) Heterozygosity for a Robertsonian translocation
 C) Heterozygosity for a non-Robertsonian translocation
 D) Heterozygosity for an inversion
 E) Two of the provided answers are correct

Handwritten notes and calculations:

$\frac{104}{200} = 0.52$
 $\frac{43}{200} = 0.215$
 $\frac{51}{200} = 0.255$
 $\frac{2}{200} = 0.01$

$TTss \times ttSS$
 $F_1 TtSs \times TtSs$
 F_2
 $\frac{104}{43} = \frac{T_S}{ttss}$
 $\frac{51}{2} = \frac{T_S}{ttss}$

You can never get more than 100%

104 all RT.
 2

37
 37
 106
 113
 200
 $9/16 \times 200 = 113$
 106
 200

14
 14
 14

17. A trans-dihybrid is created and test crossed. The following F₂ data are obtained

- Ab phenotype 53 ^{Pf}
- aB phenotype 51 ^{Pf}
- ab phenotype 47
- AB phenotype 49

You want to do the version of the Chi-Square test that takes into account viability. What value do you use for the degrees of freedom?

- A) 1
- B) 2
- C) 3
- D) 4
- E) This cannot be determined from the information provided.

18. A technique for which the separation of different molecules depends on the charge of the particular, an electric field and the fact that small molecules will travel faster.

- A) Gel electrophoresis
- B) 'Time of flight' mass spectroscopy
- C) Fluorescent activated cell sorting (FACs)
- D) Only two of the mentioned techniques match the description given.
- E) All three of the mentioned techniques match the description given.

19. Humans have many more different proteins than Drosophila because

- A) Humans have more gene splicing.
- B) Humans have more protein modification.
- C) Humans have more alternative splicing of RNAs.
- D) Only two of the reasons given are true.
- E) All of the reasons given are true.

$AABB \times aabb$
 $AA.bb \times aa.BB$
 $F_1 AaBb \times AaBb$
 $AB =$

20. To determine if a particular mRNA is expressed at a specific stage of development

- A) Use PCR with genomic DNA
- B) Use Southern blotting
- C) Use Northern blotting
- D) Use ASO
- E) Use restriction mapping

21. To determine if a primate has a gene homologous to a cloned human gene

- A) Use PCR
- B) Use Southern blotting
- C) Use Northern blotting
- D) Use ASO
- E) Use restriction mapping

22. An enzyme procedure used for detecting RFLP's

- A) Partial digestion with a restriction enzyme
- B) Complete digestion with a restriction enzyme
- C) Reverse transcription
- D) Ligation
- E) Transcription

23. A procedure used to create a probe when you know the protein's sequence

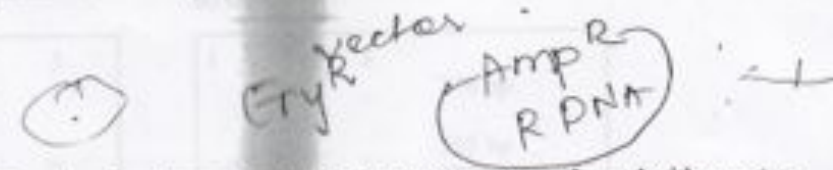
- A) ORF reading frame scanning
- B) Reverse transcription
- C) Reverse translation
- D) Antibody screening
- E) Gene tagging

RNA

Pro!

24. A plasmid vector has a gene for erythromycin resistance (Ery^R) and a gene for ampicillin resistance (Amp^R). The ampicillin gene is cut with a restriction enzyme and donor DNA, treated with the same enzyme, is combined with the vector. What genotype needs to be selected for to show evidence of transformation with recombinant DNA? Superscript * indicates sensitivity.

- A) $Amp^R Ery^R$
- B) $Amp^R Ery^S$
- C) $Amp^S Ery^R$
- D) $Amp^S Ery^S$
- E) none of the above

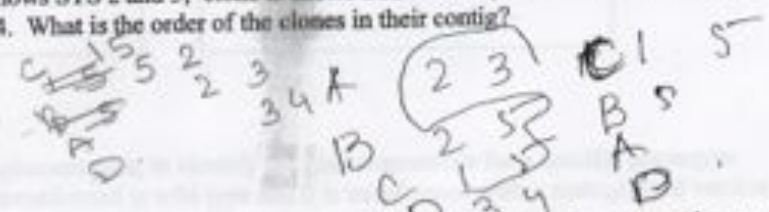


25. In the process of making a knock out mouse, embryonic stem cells are transformed with a vector carrying the tk^+ gene and a neomycin resistance gene. (The tk^+ gene converts ganciclovir into a toxic substance.) In order to screen for embryonic stem cells that have the mutagenized gene inserted at a homologous site on a mouse chromosome, you would screen for cells that were

- A) Neomycin resistant and able to grow in the presence of ganciclovir.
- B) Neomycin resistant and unable to grow in ganciclovir.
- C) Neomycin sensitive and able to grow in the presence of ganciclovir.
- D) Neomycin sensitive and unable to grow in the presence of ganciclovir.
- E) Neomycin sensitive and tk^+ .

26. Four clones (A, B, C, D) of human genomic DNA are tested for sequence-tagged sites (STS) 1 through 5. Clone A shows STS 2 and 3; clone B shows STS 2 and 5; clone C shows STS 1 and 5; clone D shows 3 and 4. What is the order of the clones in their contig?

- A) ABCD
- B) BDAC
- C) CBAD
- D) ACBD
- E) DCAB



27. In the yeast two-hybrid assay your favorite protein is being used. The bait construct is on a yeast-plasmid with the DBD exon, in a strain that has the normal $His3$ gene knocked out, and a recombinant $His3$ gene put under the control of the $gal4$ gene's promoter. In this same strain the normal, $gal4$ -specific transcription factor has been destroyed. You use this special "bait" yeast-strain to screen a yeast cell library in which the vector has a variety of ORFs bound to the coding sequence of the AD domain. To screen the library you fuse your bait yeast strain with individual library members and screen for

- A) Cells that are transcribing the $His3$ gene
- B) Cells that are transcribing the $Gal4$ gene.
- C) Cells that are not transcribing the $His3$ gene.
- D) Cells that are not transcribing the $Gal4$ gene.
- E) Cells that are not transcribing the $His3$ and the $Gal4$ genes.

GAL4 AD
GAL4 DBD
c. you get
from page

28. The first exon of the wild type beta-globin gene is cut by the restriction enzyme DdeI at the site CTNAG (where N is any base) to produce two fragments of 175 and 201 bp. The mutation causing sickle-cell anemia is a single base pair substitution where the sequence 'CTGAG' is changed to 'CTGTG'.

If the DNA from a couple and their unborn child had the beta globin gene PCR amplified and digested with Dde I to completion, which of the band profiles shown below indicates that both parents are carriers of the sickle cell trait and their fetus is homozygous for sickle cell anemia? Mom is lane 1, dad is lane 2, fetus is lane 3.

A) Gel A

	1	2	3
376 bp	-	-	
201 bp	-	-	-
175 bp	-	-	-

B) Gel b

	1	2	3	
	-	-	-	376 bp
	-	-	-	201 bp
	-	-	-	175 bp

C) Gel c

	1	2	3
376 bp	-	-	-
201 bp	-	-	
175 bp	-	-	

D) Gel d

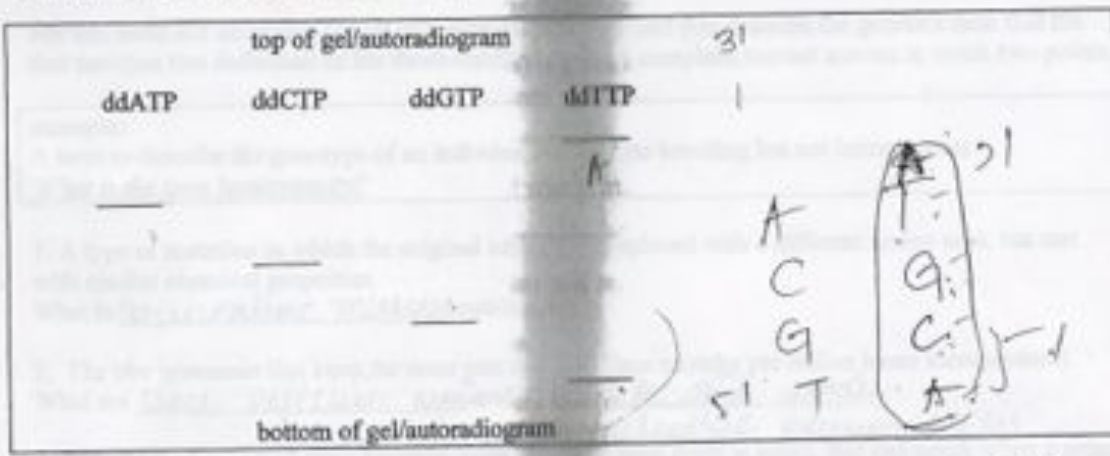
	1	2	3	
	-	-	-	376 bp
	-	-	-	201 bp
	-	-	-	175 bp

E) None of these gels

29. For functional complementation, to identify the gene responsible for a specific phenotype

- A) the organism to be transformed is wild type and it is transformed with a mutagenized version of the gene.
- B) the organism to be transformed has the mutation, and the cDNA library used for transformation is made from wild type cells that express the gene.
- C) the organism to be transformed has the mutation and the cDNA library is made from mutant cells that fail to express the gene.
- D) the organism is wild type and it is transformed with the mutant gene's promoter fused to a reporter gene.
- E) the organism has the mutation and it is transformed with the mutant gene's promoter fused to a reporter gene.

30. After using the dideox-method for sequencing DNA you observe the following



From the autoradiogram determine the base sequence of the DNA strand used as the template for the sequencing reaction. The template sequence is

- A) 3' -TACTT - 5'
- B) 5' -TGCAT - 3'
- C) 3' -ACGTT - 5'
- D) 5' -ATGCA - 3'
- E) 5' -TTGCA - 3'

3' TAGG

3' ATGCACT

5' to 3' min

Please continue on to the next section of the test

5' to 5' far

ACGTA

bottom to up.

5' to 3'

T
G
C
A

5' 3' 5'
A C G T
TGCAT
5' ACGTA 3'
3' TGCAT 5'

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 $F_1 TtSs \times TtSs$
 F_2
 $\frac{104}{43} = \frac{T_s}{ttS}$
 $\frac{51}{2}{T_S}$
 $\frac{2}{200}$

You can never get more than 100%.

104 all RT.
 2

37
 37
 106
 106
 200

14

II. Genetics Jeopardy, also known as 'Reverse definitions'.

For this section a definition (or set of conditions) is given and you describe the genetics term that fits that matches that definition or fits those conditions. Each complete, correct answer is worth two points.

example:

A term to describe the genotype of an individual who is true breeding but not homozygous.

What is the term *hemizygosity*?

1. A type of mutation in which the original amino acid replaced with a different amino acid, but one with similar chemical properties.
✓ What is *conservative missense mutation*.
2. The two processes that keep the error rate very low (one mistake per billion bases incorporated).
What are *base excision repair* and *nucleotide excision repair*.
3. The cause of improper gene function, even when the gene itself is intact, that can occur when a gene is part of an inversion or translocation.
✓ What is *position effect*.
4. An orderly visual presentation of the identification of each chromosome of the complement.
✓ What is *karyotype*.
5. A technique that uses only dideoxynucleotides, primers and polymerases.
What is *dideoxy sequencing*.
- ✓ 6. An approach to assembling sequence information that begins with the linkage map.
What is *top down approach*.
- ✓ 7. A hypothesis to explain the observation that humans share some genes only with some prokaryotes.
What is *lateral transmission of genes*.
- ✓ 8. A category of DNA that can be used for DNA fingerprinting.
What is *minisatellite*.
- ✓ 9. A technique that requires Taq polymerase and primers bracketing the DNA of interest.
What is *(PCR) Polymerase Chain Reaction*.
10. A group of cells that all contain the same recombinant DNA.
What is *DNA Clone*.

7x2 = 14

DNA
clone.

Please continue on to the next section of the test.

III. Problems, short answers and mini essay section.

1. (5 points) The cloning vector pUC19 is 2.7 kilobases and has adjacent EcoRI, HindIII and BamHI sites at the multiple cloning site [Note: the restriction sites of the multiple cloning site are so close together that for a restriction map they map to the same location.] Use the data given below to construct a restriction map for the *Bzteo* gene which has been cloned into the pUC19 plasmid.

Restriction enzyme digest	Fragment size (kilobases)
EcoRI	3.0, 2.7
HindIII	2.7, 1.8, 1.2
BamHI	2.7, 2.0, 1.0
EcoRI, HindIII	2.7, 1.8, 1.2
HindIII, BamHI	2.7, 1.8, 1.0, 0.2

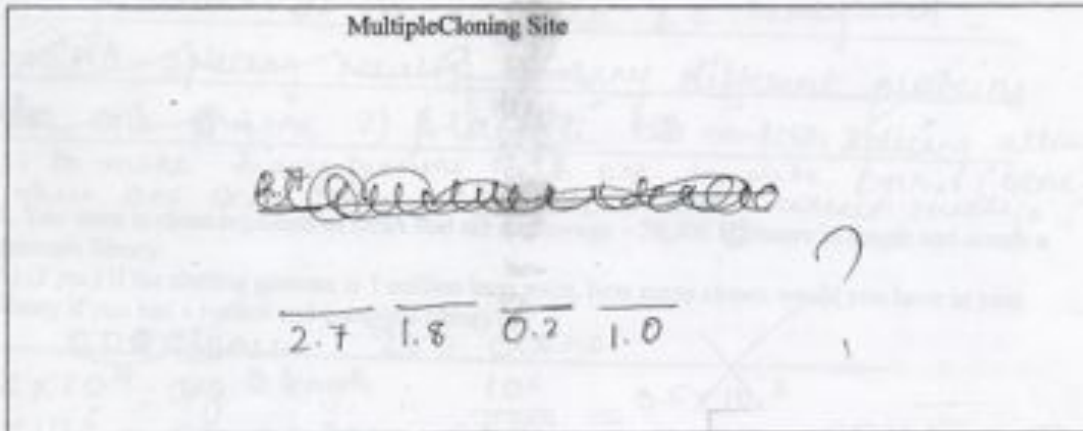
Handwritten calculations:
 $2.7 \xrightarrow{2.0} 0.7$
 $2.7 \xrightarrow{1.8} 0.9$
 $0.9 \xrightarrow{1.0} 0.2$
 $0.2 \xrightarrow{1.0} 1.0$

f) EcoRI was used to liberate the gene from the genome because

A) Which of the restriction enzymes was likely used to liberate the *Bzteo* gene from its genome and why do you think so?

~~EcoRI, BamHI, HindIII were used to liberate the gene~~
 BamHI and HindIII
 Act the gene and to release gene into restriction can be used

B) Put your restriction Restriction Map in the box provided with the Multiple cloning site in the position given.



2. (2 pts) What is the "job description" of the Genetics Material? In other words, what four properties must it have?

- It must be Replicable
- It must be Mutatable
- It must be able to direct cell function
- It must be inheritable

back

alternat splice.
DNA splice

3. (2 pts.) Two plants of the following genotypes are crossed $Aa\ bb\ Cc\ Dd\ Ee \times Aa\ Bb\ Cc\ dd\ Ee$. All five loci are sorting independently, have simple dominant/recessive allelic relationships, and there are no gene interactions and all genes are fully penetrant. What is the probability of getting an individual with one or more dominant traits? $25\% / 25\%$

1. $P(\text{getting an indiv. with no dominant trait}) = \frac{1}{4} \times \frac{1}{2} \times \frac{1}{4} \times \frac{1}{2} \times \frac{1}{4}$
 $= \frac{1}{256}$
2. $P(\text{getting indiv. with one or more dominant trait}) = 1 - \frac{1}{256} = \frac{255}{256}$

4. (4 pts.) Beadle and Tatum developed the theory one gene, one enzyme. Given what we have discussed in this course, what are all the ways that you know of that this theory is inadequate to explain the relationship of genes and gene products.

1) ~~One enzyme~~ Many of the proteins ^{require} more than one polypeptides coded by more than one gene in order to be functional as an enzyme. i.e. hemoglobin.

2) ~~RNA~~ Splicing results in many different proteins from one gene. 2) Alternate ~~the~~ m-RNA splicing allows all to make many proteins from one template DNA (gene). Thus one gene ~~one~~ polypeptide is the correct ~~the~~ life.

5. You want to clone segments of DNA that are an average $\sim 20,000$ bases in length and screen a genomic library.

A) (2 pts.) If the starting genome is 1 million base pairs, how many clones would you have in your library if you had a perfect and complete library?

~~50 clones~~ 250 clones
 $2 \times 10^4 = \text{avg } \textcircled{\text{O}} \text{ length} \therefore \frac{10^6}{2 \times 10^4} = 0.5 \times 10^2$
 $1 \times 10^6 = \text{Genome size} \therefore \frac{10^6}{2 \times 10^4} = 5 \times 10 = 50 \times 5 = 250$

B) (3 pts.) Describe how you would treat genomic DNA to generate such fragments and indicate the most appropriate vector to place it in. Be very specific.

Genomic DNA would be ~~cut~~ ^{partially} cut using a restriction enzyme. The vector DNA will also be ~~partially~~ ^{partially} cut using the same restriction enzyme. The recombinant DNA will be created by ~~ligase~~ ^{ligase} connecting vector DNA and genomic DNA via ligase. The R-DNA is ~~inserted~~ ^{inserted} into the vector.

The genomic DNA would be treated with a restriction enzyme with a recognition site of 6 base pairs. The DNA will be ~~partially~~ ^{partially} digested such that only 20% of the sites are ~~needed~~ ^{needed}. (Even with restriction site is $4^6 = 4096$)

END OF TEST I hope you have a restful and joyous holiday.

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MultipleCloning Site

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- It must be replicable
- It must be mutatable
- It must be able to direct cell function
- It must be inheritable

done

